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a patients' perspective

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Running head: Upper limb rehabilitation in FSHD

Upper limb rehabilitation in fascioscapularhumeral dystrophy (FSHD): a patients' perspective

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Highlights

- More than 80% of FSHD patients report shoulder instability which affects daily life
- Despite this, only 44.4% engaged with exercises that target the upper body
- There is a lack of evidence to inform upper-limb FSHD rehabilitation exercises
- FSHD patients support the need for further research in upper limb exercises

Key points

A web-based survey was distributed to patients on the UK FSHD registry to identify what exercise modalities people living with FSHD are undertaking in the community as a part of their ongoing rehabilitation and 2) what future research projects would gain the support of people with FSHD. Despite 85.8% of patients suffering from shoulder instability which affects daily living, only 44.4% engage in targeted upper limb exercises. Exercise selection was variable amongst FSHD patients and lack of information, pain, fatigue, availability and access to facilities, cost and time were identified as barriers to exercise.

Abstract

Objective To identify what exercise modalities people living with Facioscapulohumeral Dystrophy (FSHD) are undertaking in the community as a part of their ongoing rehabilitation and 2) what future research projects would gain the support of people with FSHD.

Design. An online questionnaire comprised of open and closed questions. Conventional content analysis was used for open questions and quantitative analysis was used for closed questions.

Setting Online questionnaire distributed to a UK FSHD registry.

Participants 232 patients on the UK FSHD registry.

Interventions None

Main Outcome Measure None

Results A response rate of 43.6% was achieved with 232 out of 532 patients completing the survey. Despite 85.8% of patients suffering from shoulder instability which affects daily living, only 44.4% engaged with exercises targeting the upper body. The themes from the data were: Understanding of disease mechanism shaping exercise choice, Lack of understanding about the condition and the benefit of exercise, Support from professionals, Barriers to exercise, and Thoughts about future research. Participants (92.2%) agreed additional research into upper limb exercises is needed and felt a 3-month arm cycling intervention with monthly clinical visits and MRI imaging would be appropriate.

Conclusions. Exercise selection was variable amongst FSHD patients and lack of information, pain, fatigue, availability and access to facilities, cost and time were identified as barriers to exercise. This may account for the limited engagement with upper-limb rehabilitation despite the high percentage of shoulder instability in patients with FSHD. Further research is needed to develop evidence-based exercise interventions and guidance for upper limb exercise prescription in FSHD and patients are supportive of this

Keywords Exercise, Joint Instability, Muscular Dystrophy, Patient Participation, Rehabilitation Upper Limb

Abbreviation

FSHD – Facioscapulohumeral dystrophy

Main text

1. Introduction

Facioscapulohumeral dystrophy (FSHD) is one of the most common inherited muscular dystrophies with an estimated prevalence in the UK general population ranging from 2.9 to 3.9/100 000¹⁻³ and affecting approximately 2400 people⁴. The overall prevalence of FSHD internationally is reportedly between 3.2 to 4.6/100 00³ but is thought to be underestimated and varies considerably by country (estimated ranges between 0.8 to 12/100 000)^{3, 5}. It is widely stated as the third most common genetic skeletal muscle disease⁶, after Duchene muscular dystrophy and myotonic dystrophy. FSHD impacts the upper extremities, torso, and also the muscles in the leg, impacting negatively on muscle mass, shoulder mobility and functional ability during tasks. This is most noticeable in functional tasks that require arm elevation above shoulder height⁷, however, functional ability at any level can be affected⁸. Activities of daily living, such as self-care e.g. combing and washing hair, reaching for objects, grasping and lifting are impaired and negatively impacts in participation and recreational activities⁸. Muscle weakness of shoulder muscles can also cause instability at the shoulder manifesting as downward translation of the humeral head with respect to the glenoid (sulcus), subluxation and recurrent dislocation which may contribute to the development of shoulder pain. Secondary features of chronic pain and fatigue also have a negative impact on quality of life^{7,9}.

For people with FSHD, evidence for informing upper limb rehabilitation is limited and does not provide appropriate guidelines for clinical implementation. Exercise prescription for rehabilitation in people with FSHD is difficult given the lack of appropriate guidelines, heterogeneity in patient symptom presentation and disease progression, and existence of co-morbidities of pain and fatigue which affect functional capacity. Despite the disease primarily affecting the upper body and associated musculature, existing studies have predominantly investigated the effect of exercise in the lower limbs for rehabilitation^{10, 11}. Moderate intensity lower limb exercises such as cycling or resistance training are reportedly safe^{12, 13} but detrimental effects are possible depending on protocol, such as intensity and load used in resistance training, and individual responses to exercise^{12, 14}. Current practice for rehabilitation of the upper limb in FSHD patients is unknown and there is no equivalent evidence to guide rehabilitation of the upper limb for both exercise type and intensity. Our clinical experience suggests that exercise is an appropriate modality for upper-limb rehabilitation FSHD patients and that current practice is variable, possibly resulting from a lack of best practice or clinical guidelines. The aims of

this study are therefore to identify 1) what exercise modalities people living with FSHD are undertaking in the community and 2) what future research projects would gain the support of patients with FSHD and would be manageable for them.

2. Materials and methods

Research and Development approval for this work has been received from The Robert Jones and Agnes Hunt Orthopaedic Hospital NHS Foundation Trust. No ethical approval was needed as per the principles set put in NIHR involve. All participants were fully informed about the study, the voluntary nature of their participation, and informed of their ability to withdraw data at any point prior to them taking part in the study. As this was not a clinical trial there is no clinical trial registration number. A web-based survey developed in SurveyMonkey Audience¹⁵ (Appendix 1) was distributed electronically using the UK disease specific FSHD registry¹⁶. Responses were collected between March and June 2015 during which people received an invite to the study and two subsequent reminders. The questionnaire was developed by a team of clinicians (Specialist neuromuscular medical consultants and physiotherapists) who routinely work with people with FSHD and had nine questions, with a mix of open (n=2) and closed (n=7) questions. The two open questions asked participants to describe 1) which exercises they considered feasible for their condition and 2) the specific exercises they do for their upper body. The seven closed questions were related to patients' experiences of instability, exercise and opinions about future research. Only secondary, anonymised data were analysed, conventional content analysis was conducted according to the stages outlined by Hsieh and Shannon¹⁷. Codes were generated by a single author (non-clinical) and were then verified with another author (clinical) before being used to construct themes according to prevalence and relevance to the research questions.

3. Results

Results for response rate to questionnaire

In total, 232 participants completed the survey out of 532 who were invited (43.6%). The average response rate of the fixed answer questions was 99.4%. An overview of the fixed answer responses has been provided in table 1. Of the 232 who completed the survey, 193 participants (83.2%) answered the first open ended question, and 124 participants (53.4%) answered the second. The responses to the open-ended questions were mostly one word to single sentences, with some longer multiple sentence responses.

Results for responses related to exercise

The survey results identified that 85.8% of participants agreed that shoulder instability affects their daily life. Despite this, only 44.4% of the total population reported engaging with exercises that target the upper body. Of these, 32.3% of respondents performed exercises which targeted their upper body more than once a week and the remaining 12.1% performed exercises targeting the upper body less than once a week. Of those who do exercise, there were a wide range of activities and exercises described by participants summarised in table 2.

Most long-answer responses included some exercises or activities that the participant carried out, with some individuals also describing the positive impact that it has on their condition, for example in this statement about arm cycling:

“I already do arm cycling at nmc and I can tell the difference with movement and less aches”

There were very few who described negative repercussions from exercise, and where these were present, they were often related to a specific exercise or piece of equipment.

“I struggle using a cross trainer due to my FSHDM as the arm movements lead to cramps in my trapezius muscle and severe pain and swelling in both collar bones!”

These participants' experiences suggest that exercise offers a beneficial mechanism to improve flexibility and strength and that a wide range of exercises can be utilised to target different locations in the body, including the upper limbs. Responses vary between submissions, suggesting that these exercises help participants differently according to their symptoms.

Following the application of thematic principles, the following themes were constructed from the data

1. Understanding of condition shaping exercise choice
2. Lack of understanding about the condition and how exercise interacts with it
3. Support from professionals
4. Barriers to exercise
5. Thoughts about future research

Understanding of condition shaping exercise choice

Participants were knowledgeable of the importance of exercise to prevent the deterioration associated with the condition, emphasising the knowledge and understanding that they held in this area. This desire to work in order to counter the effects of FSHD, or acquire a greater understanding of the condition and use that knowledge to determine useful exercises, is common in the survey responses. Listed exercises were often accompanied with explanations of how it interacts with their condition e.g.

“1. Pliates to improve core muscle strength 2. Yoga to maintain flexibility, and to enable arm cycling while braced on the floor 3. Walking whilst possible”

Many responses in the survey mention exercises or equipment that participants have tried, or previous research studies that they have engaged with, with varying levels of success. Participants reported using this accumulated knowledge to assess the potential suitability of exercises or machines. This extends to items that have already been used with problematic results and there seemed to be a willingness to try again in case a mistake was made or the item has been adjusted in some way:

“I have tried to use that piece of equipment several times over the last decade, unfortunately each time it has caused severe pain and breathing difficulties each and every time. Having said that it has never taken place under supervision, so I think with the correct set of circumstances it is definitely worth a trial.”

Lack of understanding about the condition and the benefit of exercise

Of the participants who do not engage with targeted upper limb exercise, two main groups were observed. A minority of participants (<5%) did not participate in specific or additional exercise outside of daily activities/occupational requirements due to a perception that exercise was not beneficial, for example,

“do no specific exercise, [finding] regular movement during the day [...] is of much benefit to the maintenance of my remaining shoulder movement”,

Approximately 10% of participants reported not exercising due to confusion surrounding the benefits of exercises or a lack of knowledge about their condition. These views were captured by the following comments:

“Unsure, as have never had any correct exercises suggested.”

“I would definitely benefit from arm exercise but do not know what type would be possible.”

The impact on wellbeing and quality of life is presented in some of the survey responses with participants reflecting on their lack of knowledge and the impact that it could have had on their life had they known about it, for example:

“Not sure but wished I knew more on what i can and cant do [sic]”

“I haven’t done any exercises at all over the years because I accepted my condition and didn’t realise that exercise would help...”

Some participants showed concern about the availability and quality of information which might help an individual with FSHD and were worried that this was leading to inconsistent advice and exercise recommendations.

Support from professionals

Whilst treating clinicians offered exercise advice, this was described as lacking consistency in some cases. A notable number of participants mentioned advice and recommendations that they received from professionals/instructors in community settings or alternate professions, which they use to guide their exercises, for example, Pilates instructors, gym instructors, personal trainers, and osteopaths. This was often coupled with an explanation of how the action benefits their strength or mobility, for example:

“Working with a level 4 gym instructor, we have been targeting exercises to increase movement of arms to raise to shoulder level and above. We have been trying to isolate specific muscle groups and working to strengthen these.”

Barriers to exercise

Lack of knowledge was not the only element that contributed to participants not exercising. In the survey, several other factors or barriers were identified which influence people's propensity to exercise, namely loss of mobility, reduced functional and travelling ability, pain and fatigue. Discomfort (primarily pain) hindered the ability of patients to engage with specific exercises.

Functional limitations associated with the condition of FSHD were the most common barrier raised. Many participants in the survey noted that their condition affected their travelling ability, limiting their ability to engage with exercise regimes or research studies, saying for example:

"I am unable to travel now, and would be unable to take part, other than to give my comments as above."

These limitations are frequently identified in the survey responses, with many participants offering stipulations in their assessment of exercises, such as "doesn't involve", "limited in what I'm able to do" or "progression of disease made it impossible", "used to do this until".

Pain from exercise or problems with exercises was a significant barrier discussed in the survey responses. Several participants described situations which exercises or machines which "caused severe pain and breathing difficulties each and every time", some of which resulting in "[having] to refrain from even trying for the last 4 months." However, patients were able to implement a range of compensatory strategies (e.g. exercising parts of the body not affected by pain or taking a temporary break from a specific exercise. Although problematic, this was not always a complete barrier, as many of the responses reported continuing to exercise areas of their body which were pain-free.

In addition to the condition-based barriers, the survey responses mentioned availability and access to facilities, whether through cost, location or time, as a barrier contributing to lack of exercise. Although many of these barriers raised cannot be easily overcome, some individuals offered some suggestions of ways to improve exercise engagement.

Thoughts about future research

Most participants (92.2%) agreed that research into upper limb exercises for people living with FSHD was needed and that engagement with clinical trials would not be a problem if duration was no more than 3-months (73.7%) and subsistence costs were reimbursed. Patients stated that an arm cycling intervention could be a priority (56.9% agreed and 38.8% unsure) and were willing to have an MRI scan (76.4%).

4. Discussion

The aims of this study were to identify 1) what exercise modalities people living with FSHD are undertaking in the community and 2) what future research projects would gain the support of patients with FSHD and would be manageable for them. We achieved the aims of our study, having identified the range of exercise modalities undertaken by people with FSHD and proportion of participants who use exercise as a part of their upper limb rehabilitation. We were also able to identify participants' perceptions of upper limb exercises and associated barriers for participation which can be used for informing management and future research.

There is insufficient evidence to inform clinical decision-making regarding exercise in FSHD and this is likely limiting clinicians' ability to prescribe exercise and the ability of people with FSHD to engage with it. Within our study it was identified that more than 50% of the respondents were not engaging in upper-limb exercises, despite more than 80% of participants reporting shoulder instability which affects their activities of daily living. Shoulder instability is a complex phenomenon with poorly understood mechanisms¹⁸. This is particularly true for people with FSHD where the mechanism for shoulder instability may be different as a result of the changes to either the biomechanics (e.g. properties of the musculoskeletal architecture/tendon) or the neural control (e.g. recruitment of motor units) of the musculoskeletal system associated with the condition¹⁹. The aforementioned impairments relating to biomechanics and neural control may be assessed using imaging such as MRI and 3D movement analysis in combination with electromyography respectively. Responses of people with FSHD were supportive of MRI which may provide further insight into mechanisms of shoulder instability in FSHD and inform upper-limb rehabilitation¹⁸. Of the people who exercised, a broad range of exercise modalities were identified, and many participants suggested that they also seek support outside of their immediate care team. Whilst variability in exercise selection may be reflective of the heterogeneity within this patient group, our study

suggests that the variability may reflect the lack of evidence-based guidelines for FSHD and how exercise interacts with it.

It is important to identify a safe range of exercise modalities and parameters that can be carried out by people with FSHD. Existing research studies had focused predominantly on the lower-limbs^{1, 4, 6, 11-13, 20-22} in which some exercise programmes have demonstrated effectiveness, for increasing cardiovascular fitness in FSHD^{23, 24} and other neuromuscular disorders^{20, 21}, though others have suggested that some exercise intensities can be detrimental^{12, 14}. There are also other secondary benefits associated with physical activity such as decreased risk of comorbidities and improved overall wellbeing²⁵. It is possible that the rate of functional decline in people with FSHD who do not engage appropriately with exercise is faster relative to those who do for both the upper and lower limbs. Further research is needed to confirm this, however, based on the findings from existing lower limb research, it is reasonable to assume that similar benefits may be achieved for the upper limbs²² and patients' views confirm this:

“I already do arm cycling at [specialist centre] and I can tell the difference with movement and less aches”

Further research focusing specifically on upper limb exercises for people living with FSHD is therefore needed. It is important that when developing or investigating exercise interventions in FSHD, that these have appropriate physiological rationale and sufficiently capture the benefits of exercise. Exercise interventions should aim to address the main mechanisms associated with the disease pathophysiology or subsequent symptoms, and consider the limitations associated with the condition to allow it to be delivered safely and effectively.

A series of barriers and facilitators to people with FSHD exercising were presented in the data, highlighting a series of key elements which must be considered when developing future research. Knowledge of the effects of exercise were drivers for participation and a lack of understanding was a barrier, therefore, in addition to quantifying the benefits from exercise research should explain physiological mechanisms of action and risk of harm. For example, strengthening exercises may not result in hypertrophy but are likely to have positive secondary effects on psychological wellbeing²² and may limit the rate of functional limitations associated with the condition²². Some of the barriers identified were similar to those experienced in the general population e.g. pain associated with exercise, and it is not clear if these events are indicative of normal or detrimental responses

to exercise. It is important that professionals engaged in exercise prescription are routinely trained in the state-of-the-art knowledge in terms of benefits and risks associated with exercise to improve consistency in the messages being given to patients^{10, 13}. This is particularly important given the degenerative and long-term nature of the condition, as patients with FSHD will be reviewed by professionals throughout the course of their condition and be required to self-manage.

Secondary complications associated with the condition, namely pain, though loss of mobility, reduced functional ability, and fatigue were also mentioned by participants as barriers to upper limb exercise. These findings are consistent with other research studies investigating the natural history of people living with FSHD and exercise²². In addition to the condition-based barriers, the survey responses mentioned availability and access to facilities, whether through cost, location or time, as a significant barrier contributing to lack of exercise. Limited evidence, investigating barriers to exercise for people living with FSHD in the community is available. It is therefore important that when designing exercise interventions for future studies that these factors are taken into consideration. For example, arm cycling has the potential to be an effective, low impact exercise, however if required to travel to use the necessary equipment, this could limit access and participation.

Study Limitations

Within our study we achieved a response rate of 43.6% for the questionnaire. This is less than half of all people in the UK FSHD registry, however, it still captures a meaningful proportion of the true FSHD population. While other studies may have higher responses rates and number of responders, this is reflective of a sample rather than the known population and so it is important to interpret the response rate against this understanding. It is recognised that our study could potentially underestimate the number of people who are not undertaking upper limb exercises given that people with FSHD who do not exercise 1) may be less informed and likely to engage with research investigating modalities that do not feature in their routine management of the condition and 2) may have a lower functional capacity, possibly stemming from the disease progression and limited engagement with exercise, which negatively affects their ability to engage with the questionnaire.. Further work may therefore be required to identify exercise interventions for lower functioning patients or methods for engaging people with FSHD not currently doing exercise. A wider range of methods of data collection e.g. Qualitative research methodologies or use of paper questionnaires, would also be beneficial for capturing information

regarding patients experiences of exercise/rehabilitation and avoid loss of engagement due to factors like reduced digital access²⁶.

Conclusions

Overall, participants were positive and agreed about the need for future research into exercises for people living with FSHD. The responses indicate that people with FSHD are willing and are able to engage in upper limb exercises. Further research is therefore needed to develop appropriate, evidence based exercise interventions and guidance for informing exercise prescription for upper limb rehabilitation in FSHD. In absence of pharmacotherapy or gene therapy, upper limb rehabilitation is fundamental in the ongoing management of FSHD. The results of our study provide support for the use of a 3-month arm cycling intervention, performed at home or in the clinic, as well as the use of MRI and frequency of clinical visits. The barriers and recommendations identified by participants will also be important for designing research studies, as mobility, pain, access, motivation and fatigue were all identified as factors which may negatively impact on research design, recruitment and overall translation into clinical practice.

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Table 1. Overview of responses to the fixed answer questions

Q1. Do you think there is need of research on exercises for upper limbs in your condition?		
	<i>Frequency</i>	<i>%</i>
Yes	214	92.2
Not sure	13	5.6
No	4	1.7
Incomplete	1	0.4
Q2. Does shoulder instability impact on your daily life?		
	<i>Frequency</i>	<i>%</i>
Agree	199	85.8
Not sure	15	6.5
Disagree	15	6.5
Incomplete	3	1.3
Q3. Do you think arm cycling as a feasible exercise in your condition?		
	<i>Frequency</i>	<i>%</i>
Agree	132	56.9
Not sure	90	38.8
Disagree	9	3.9
Incomplete	1	0.4
Q4. What do you think about duration of home exercise programme of arm cycling for 3 months?		
	<i>Frequency</i>	<i>%</i>
It is sufficient	171	73.7
No comments	51	22.0
It is too long, should be shorter	8	3.4
Incomplete	2	0.9
Q5. This study will need participants to visit study centre for assessment of strength and function of upper limbs. Participants will attend at 4 weekly intervals. What is your opinion about visits?		
	<i>Frequency</i>	<i>%</i>
No change	83	35.8
Would attend all visits if travelling expenses are paid	134	57.8
Need fewer visits	15	6.5
Incomplete	0	0.0
Q6. This study would also involve having MRI scan before and after study period. An MRI scan would take about 45 minutes. What is your opinion about MRI scan?		
	<i>Frequency</i>	<i>%</i>
Yes, it is possible	173	74.6
It would be uncomfortable to lie on scanner for 45 minutes	40	17.2
No comment	19	8.2
Incomplete	0	0.0
Q7. Do you currently do specific exercise targeting your upper body?		
	<i>Frequency</i>	<i>%</i>
Yes, more than once per week	75	32.3
Yes, less than once per week	28	12.1
No	127	54.7
Incomplete	2	0.9

Table 2: Summary of exercises reported by people with FSHD.

General physical activity – no additional exercises outside of daily activities/occupational requirements	Regular movement, no specific exercise, general walking, stair walking, daily tasks/everyday activities
Resistance training with load or assistive machines	Rubber band/resistance band exercises, low impact- low weight exercises, strengthening exercises, weights, weights for arms, shoulder rope pulleys/ shoulder machines, Ab cruncher, Gym workout/ programme, cable multi-gym
Water-based	Hydrotherapy/pool based exercises, swimming, Aquafit
Cardiovascular training	Bike riding, gentle rowing, cross trainer, punch bag, arm cycling
Body weight exercises	T'ai chi Pilates, Plank, Floor exercises, Wall push ups/ push ups
Maintaining range, mobilising or stretching	Yoga, Qigong, neck exercises, stretching, Slow stretches, door based exercises, rotation
Core exercises	Pilates, Floor exercises
Therapist assisted	Physiotherapy, Osteopathy
Other/ non descriptive	Arm exercises, palm to palm exercises, deep breathing, adapted game based exercises e.g. throwing and catching, wii sports